PATIENT INFORMATION

Genetic carrier screening

What is genetic carrier screening?

Reproductive genetic carrier screening (sometimes referred to as genetic carrier screening, carrier screening, or expanded carrier screening) involves analysing genes from an individual's blood or saliva sample to identify carriers of certain genetic changes that can increase the chance of having a child with health conditions.

What are genes?

Genes are short sections of DNA that provide specific instructions for our body to grow, develop and function. An individual's genetic information is then packaged into tiny threadlike structures called chromosomes. A typical human cell contains 46 chromosomes, or 23 chromosome pairs (with one copy from each pair usually originating from the egg, and the other copy originating from the sperm). These chromosome pairs are labelled 1 to 22 (the autosomes) and X and Y (the sex chromosomes). We all carry variations in our genetic code, which makes each of us unique. However, some gene variations (also known as mutations) can disrupt how a gene works and cause health implications. Individuals with these variations may experience health implications themselves or be at risk of having children with a genetic condition.

Should I have reproductive carrier screening?

The decision to have screening is a personal choice made by the individual or couple. It is not mandatory. However, it is important individuals are aware this testing is available for them. Your fertility specialist or physician may have suggested you consider reproductive genetic carrier screening. The Royal Australian and New Zealand College of Obstetricians and Gynecologists (RANZCOG) advise that couples planning to conceive or in early pregnancy should be offered genetic carrier screening as part of their reproductive planning and care.

It can often be helpful to do research and speak to a genetic counsellor to help make this decision. You can learn more about genetic carrier screening by reading this information and exploring the carrier screening guide by the Reproductive Genetics team at Victorian Clinical Genetics Service (VCGS) here: https://carrier-screen-guide.vcgs.org.au/.

What sort of genetic conditions are included on genetic carrier screening?

The two major types of inheritance that can lead to a healthy couple having children with serious genetic conditions are called autosomal recessive and X-linked recessive.

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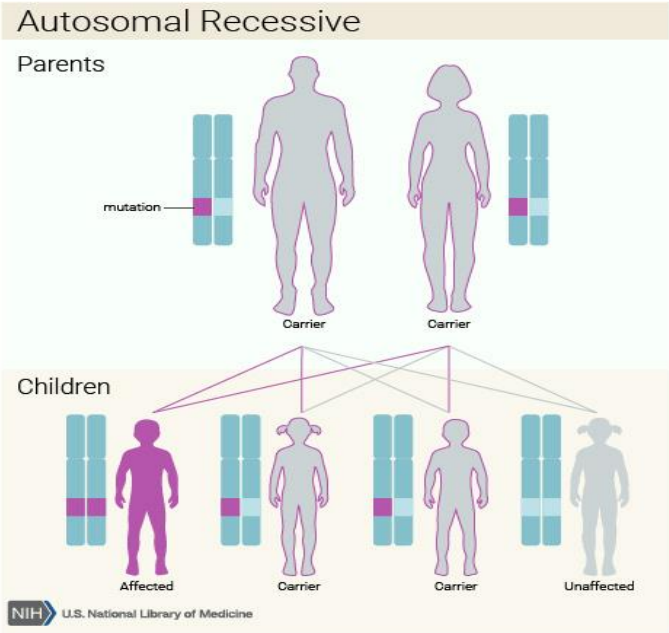
Autosomal recessive conditions

An autosomal recessive condition means both copies of the gene must be faulty for the condition or trait to develop. Carriers of recessive conditions are usually healthy and have no signs or symptoms associated with the condition. They are therefore sometimes referred to as 'silent carriers'. Common examples of recessive conditions are Thalassemia and Cystic Fibrosis.



If both individuals within a couple are found to be carriers of the same recessive genetic change/condition, each of their conceptions will have a 25% (1 in 4) chance of inheriting the condition.

X-linked conditions

X-linked conditions are caused by genetic changes occurring on the X chromosome. Males typically have a X and a Y chromosome in each cell whilst females usually have two X chromosomes. When there is a genetic change on the X chromosome, females are generally unaffected or less severely affected than males as they have a second unaffected copy of the gene on their other X chromosome. Males are therefore generally more severely affected by X-linked conditions as they typically only have one X chromosome (i.e. if there is a genetic change on their X chromosome they do not have a second unaffected copy to compensate).

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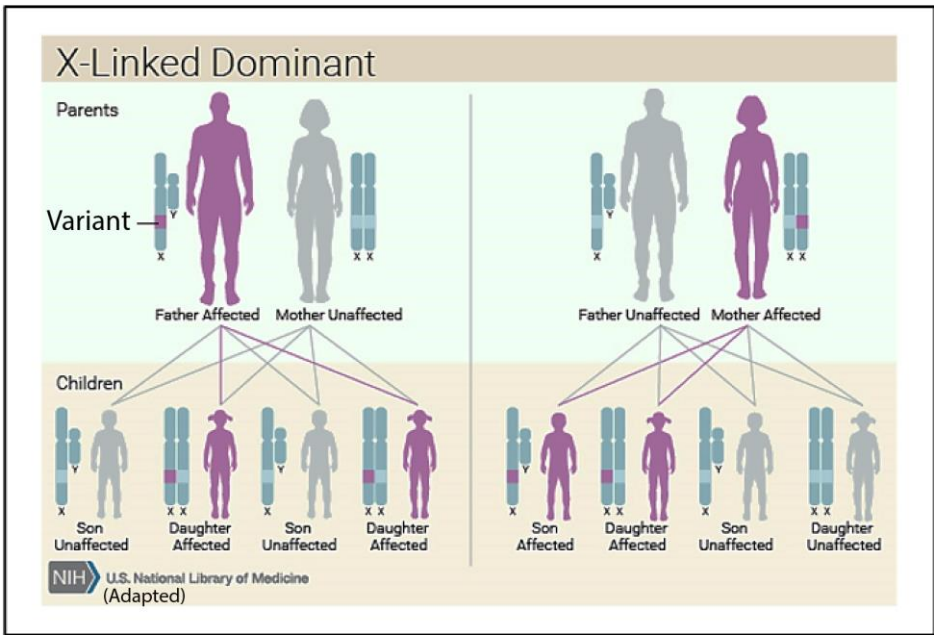

If a female is a carrier for an X-linked condition, there will be a 1 in 4 chance of having a son affected by the condition for each conception. The most common X-linked recessive condition is Fragile X syndrome.

How do I access reproductive carrier screening?

There are many laboratory and genetics services that offer different forms of carrier screening. No1 Fertility recommends genetic carrier screening through the Reproductive Genetics team at Victorian Clinical Genetics Services (VCGS) via the two options below:

prepair (https://www.vcgs.org.au/tests/prepair): This test screens for 3 conditions that are commonly carried in the population (Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X Syndrome). Currently this test costs $389, and the egg contributor is tested first. If they are found to be a carrier of one of these three conditions, testing for the sperm contributor is then arranged and is bulk billed.

prepair+ (https://www.vcgs.org.au/expandedcarrier): This tests screens for about 300 common and rare conditions, including the three conditions in the *prepair* test. Testing of the egg and sperm contributor occurs at the same time. Currently the cost for this test is $900 for both individuals to be tested. Sometimes these tests are called expanded carrier screening.

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To arrange genetic carrier screening, please contact the Reproductive Genetics team at VCGS on 9936 6402 or at screeninggc@vcgs.org.au and inform them you are a patient of No1 Fertility.

What if I/we have no family history of a genetic condition?

Approximately 90% of individuals who are carriers of a genetic condition, do not have a family history of the genetic condition(s). Individuals without a family history of a genetic condition should still consider carrier screening.

What does it mean if I or my reproductive partner are 'carrier/s'?

It is common to be a carrier of at least one genetic condition. Carriers are usually healthy and do not have any symptoms, which is why most carriers are unaware. However, it is important for your partner to consider screening to determine if they are also a carrier of the same condition, to determine if there is risk to future offspring. Where both members of a couple are found to be carriers of an autosomal recessive condition or a woman is found to be a carrier of an X-linked condition, there is a 1 in 4 chance of each pregnancy being affected by the condition. When you are found to be a carrier of a recessive genetic condition, your first-degree relatives (your siblings, parents and children) are at a 50% chance of carrying the same genetic change and might benefit from genetic counselling to discuss testing in themselves.

What does it mean if my reproductive partner and I are at 'increased risk'?

Individuals or couples found to be at risk of having a child with a genetic condition can choose to conceive naturally, or through IVF. If they choose to conceive naturally, there are specific tests during the pregnancy that can determine if the pregnancy has inherited the genetic condition. If they choose to have IVF treatment and create embryos, often a test can be developed for testing embryos via preimplantation genetic testing for monogenic conditions (please see *Patient Information: PGT-M* for further information) to determine if they are suitable for transfer (unaffected or some carriers), to reduce the risk. Both pathways are very personal decisions and can be discussed in detail with your Fertility Specialist or Genetic Counsellor.

Can I start my IVF treatment with my carrier screening results pending?

Yes. However, you need to be aware there is always a small chance you will be found to be at an increased risk of having a child with a genetic condition. For individuals undertaking expanded carrier screening (such as *prepair+*) there is approximately a 2% chance they are found to be at increased reproductive risk.

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What is the cost?

The cost of carrier screening depends on the type you choose. The two options offered by VCGS are:

Core panel - *prepair* (https://www.vcgs.org.au/tests/prepair): This test screens for 3 conditions that are commonly carried in the population: Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X Syndrome. Currently this test costs $389, and the egg contributor has testing first. If they are found to be a carrier of a condition, then testing for the sperm contributor is bulk billed.

Expanded panel - *prepair+* (https://www.vcgs.org.au/expandedcarrier): This tests screens for about 300 common and rare conditions, including the three conditions included in *prepair*. Testing of the egg and sperm contributor occurs at the same time. Currently the cost for this test is $900 for both individuals to be screened.

How long do results take?

Results for *prepair* (core panel) take approximately 2-3 weeks to be reported, and results for *prepair+* (extended panel) take approximately 4-6 weeks to be reported. You will be contacted to discuss your results for testing arranged through the VCGS Reproductive Genetics team and linked with your care at No1 Fertility.

Questions?

If you have read this information and have questions, please contact the Nº1 Genetics Department at 9132 9600 or genetics@number1fertility.com to speak to one of our Genetic Counsellors.

To arrange genetic carrier screening, please contact the Reproductive Genetics team at VCGS on 9936 6402 or at screeninggc@vcgs.org.au and inform them you are a patient of No1 Fertility.

The information provided above is intended for educational purposes only and should not be used as a substitute or replacement for medical advice received from a medical professional. It is important to discuss your individual circumstances and situation with your treating doctor.

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